2026 Texas Focus:

Part I. Down Syndrome and CVI: an Overview

Friday June 5, 2026 I 8:30-9:45 am

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Down syndrome overview:

- **What is it?**
  - Most common chromosomal condition, affecting 1 in every 640 babies born in the US.
  - Characteristic features: shorter stature, shorter arms/legs, flatter nasal bridge, eyelid creases with almond shaped eyes, lower muscle tone
  - More alike than unlike their family members
  - Development occurs in typical sequence, tends to be slower
  - Mild to moderate cognitive delays most common
  - Wide developmental ranges

What causes it?

- Genetics: **"Trisomy 21,"** three (not two) of chromosome 21
  - Full trisomy 21
  - Translocation – piece of chromosome 21
  - Mosaicism – only some cells contain trisomy 21
    - Extra genetic material affects how the baby develops

- To recognize and celebrate the Down syndrome community, World Down syndrome Day is **3-21** every year!

Outlook for individuals with DS today is better than ever

- Robust, energetic, collaborative DS community
- Advances in research, health care, education
- Improved life expectancy, quality of life, opportunities

The "Down syndrome Phenotype"

- Research-based description of strengths and challenges for individuals with DS, helps us anticipate challenges and use strengths to create appropriate supports for learning and development.
  - Medical research - describes the medical conditions that are common in DS (and those that are not): include lower muscle tone, ligamentous laxity, health conditions like congenital heart conditions, thyroid disease, vision and hearing challenges.
  - Educational research describes the typical learning style of people with DS – like visual learning strengths, verbal memory challenges; receptive language tends to be stronger than expressive.
  - Developmental research describes common social tendencies of people with DS: relational connectedness tends to be a strength, with fear of failure and acting out or "playing cute" when faced with a challenge; often excellent imitation skills – facial expressions, gestures, dance moves.
  - Behavioral and mental health research describes higher prevalence of autism/ASD, ADHD, mental health issues (anxiety, depression), Down Syndrome Regression Disorder (DSRD); difficulty with sense of time may make past events feel very present, memories may replay like a movie; may use

tactics to avoid hard tasks – fear of failure, distraction, elopement, hiding, “drop and flop”

- Developmental milestones:
  Wide range of achievement accepted (but are we missing additional factors affecting development that need to be addressed?)

When the expected developmental pattern doesn’t fit, what do we consider?

- Families and clinicians may notice:
  - Less eye contact than expected
  - Withdrawal in busy environments
  - Repetitive behaviors
  - Delayed communication
  - Avoidance of certain tasks

Possible reasons we explore:

- Autism spectrum disorder
- ADHD or attention differences
- Anxiety
- Medical factors
- Hearing of vision differences
- Sensory overload

Note: the same behavior can have different causes

- We often don’t consider the significant impact of brain-based blindness (CVI) or of hearing challenges in DS, but together they constitute “the perfect storm” of functional deafblindness

Resources: Down Syndrome

1. National Down Syndrome Congress
2. National Down Syndrome Society
3. Global Down Syndrome Foundation
4. Down Syndrome Diagnosis Network
5. Lumind IDSC (now merged with NDSC research)
6. Advocate Medical Group Adult Down Syndrome Center
7. Down Syndrome Education International
8. GiGi's Playhouse
9. Down Syndrome Clinic to You (DSC2U)

Visual Impairment in Down syndrome

How do we see?

- Eye – focuses image on back of brain, occipital lobe
- Brain – processes the image, to understand it, retrieve it for use
- Individuals with Down syndrome are at risk for both eye and brain related visual impairment

Jain, A., Boyd, N. K., Paulsen, K. C., Vogel, B. N., Nguyen, L., & Santoro, J. D. (2023). Ophthalmologic and neuro-ophthalmologic findings in children with Down syndrome. American Journal of Medical Genetics Part C: Seminars in Medical Genetics, 193C:e32068. https://doi.org/10.1002/ajmg.c.32068

Common eye conditions in DS (60-80% affected) – treated with glasses or surgery:

- Decreased visual acuity
- Accommodative, refractive errors
- Strabismus
- Nystagmus
- Disorders of cornea (ie keratoconus)
- Disorders of the iris

- Disorders of the lens (cataracts)
- Disorders of the retina and optic nerve
- Glaucoma
- Blepharitis
- Nasolacrimal duct obstruction

Cerebral/Cortical Visual Impairment (CVI): A Brain-Based Visual Processing Disorder – requires visual accommodations and supports:

- Ocular condition(s) alone does not explain the functional vision impairment
- Visual acuity may be normal
- Eyes may be normal
- But the child functions as someone who can't see
- If we understand that you "see" in the brain, then this can help us better understand what CVI is
- Because CVI is a brain-based visual impairment, the visual challenges can fluctuate with things affecting the nervous system (anxiety, fatigue, multiple sensory inputs)
- Not static like an eye-based condition
- CVI can be hidden or disguised by behaviors attributed to other conditions (like Down syndrome itself)

When to suspect CVI – when someone with DS:

- Looks at things differently
- Has trouble finding or recognizing things
- Has difficulty with seeing things in the distance or moving quickly
- Has challenges with hand-eye or eye-foot coordination
- Needs help navigating spaces
- Struggles in busy, noisy spaces
- Has trouble interacting with people

- Gets visually tired easily
- Struggles with school tasks
- Inconsistency with skills

Resources: CVI Now Downloadable One-Pagers (CVI and related conditions):

What are risks for CVI in Down syndrome?

i. **Any injury to the brain can cause CVI**
   - Before birth
   - At the time of birth
   - Or after birth

ii. **This includes medical issues that commonly affect babies with Down syndrome:**
   - Low oxygen levels (may need blow by oxygen or a mask for a short time)
   - Low blood sugar (poor feeding)
   - High bilirubin (jaundice; due to low fluids from poor feeding, treated with bili lights or blanket)

- We are not yet sure if Down syndrome itself is a risk for CVI, or whether Trisomy 21 makes babies more sensitive to the medical issues listed above.
- We know the incidence of CVI in DS appears to be somewhere between 38% and 50%

CVI in DS: What does it look like?

- Published literature:
  - CVI behaviors are common in Down syndrome – Woodhouse (2021)
    - 38% met behavioral threshold for "CVI suspect" needing full functional visual evaluation
  - **Reference:**
  - Wilton GJ, Woodhouse R, Vinuela-Navarro V, England R and Woodhouse JM (2021) Behavioural Features of Cerebral Visual Impairment Are Common in Children With Down Syndrome. Front. Hum. Neurosci. 15:673342. doi: 10.3389/fnhum.2021.673342

Case series describing 3 teens with DS+CVI demonstrated:

- Eye conditions common in DS present in all three
- Delayed developmental milestones, particularly skills requiring vision
- Language challenges (more preserved if ASL and multimodal communication techniques continued from infancy)
- Behaviors common in all three (anxiety with transitions, startle with loud noises, distractibility, visual behaviors)
- Late diagnoses at 15 yrs (two of the three), re-evaluation and confirmation at 19 yrs (third case)
- All three teens are responding to CVI accommodations/deafblind techniques

- **Reference:**
- Boatwright E, Banihani R, Willems I, Lehman K, Mazel E, Mark H, Wong M, Vietzman S, Chandna A and Heidary G (2025) Cerebral/Cortical visual impairment (CVI) in Down syndrome: a case series. Front. Hum. Neurosci. 19:1563420. doi: 10.3389/fnhum.2025.1563420

Functional Deafblindness: a Perfect Storm

- Up to 90% of individuals with DS have hearing challenges
    - fluctuating hearing due to fluid or infection or wax;
    - permanent hearing loss due to conductive or sensorineural hearing issues
- Even mild hearing loss with CVI constitutes deafblindness, where impact on learning and development is exponential rather than additive
- Deafblindness is significantly underdiagnosed

Examples of CVI and Dual Sensory Impact (deafblindness) in Down syndrome

- **Visual Function** – can vary depending on:

    - External Environment
        - Noise
        - Movement
        - Crowding
        - Noise

    - Internal Environment
        - Stress
        - Fatigue
        - Illness

    - Can result in behaviors that may be misinterpreted
        - Difficulty with distance viewing
        - Difficulty distinguishing objects in crowding

        - Missing animal in zoo but seeing the fence, until animal moved behind a clear partition

- **Navigation, Motor Skills**

    - Challenges with:
        - Uneven terrain
        - Crossing thresholds
        - Curbs
        - Stairs

- **Language and Communication**

    - Lack of visual attention can lead to missed incidental learning, which affects:
        - Concept development
        - Vocabulary
        - Communication
        - Facial expressions
        - Social interaction

    - Especially challenging if hearing impacted as well: sound and its source may not be meaningfully connected

Behavior in DS+CVI/DB

- **Reactive**: response to unexpected sounds or events or stress related to not knowing what is happening or what will come next
    - Grabbing
    - Dropping/grounding
    - Anxiety with transitions/refusal
    - Eloping/hiding

- **Compensatory:** may help someone with CVI get the help they need to visually access the world

    - Clearing tabletop surface
    - Dumping bins/containers
    - Looking away to listen or touch
    - Moving or spinning
    - Commenting or asking questions (may help locate a person, or switch sensory mode to auditory)
    - Relying more heavily on tactile sense

- **Visual and Auditory Fatigue – may result when:**

    - The environment is complex
    - There are too many sensory inputs at once
    - This can lead to meltdowns, frustrations, and shutting down

Visual Access and CVI

- When seeing is hard, learning and behavior can look different
- When visual access improves, abilities often become easier to see
- Visual strategies can be employed to improve access
- Behavior may change when access changes
- Learning and development may change when access changes

Resources – Down syndrome and CVI:

- Down Syndrome Center Podcast: Central Auditory Processing Disorder (CAPD) and Cerebral/Cortical Visual Impairment (CVI) in Down syndrome

- Video and Article: Down Syndrome and CVI Explained (Perkins CVI Center)
- Article: www.asharedvision.org/uploads/8/8/2/7/88271548/talking_the_language_of_the_hands_to_the_hands.pdf?from=hub

Resources for CVI, Dual Sensory Impact:

1. **Texas School for the Blind and Visually Impaired**
2. **CVI Now, Perkins School for the Blind**
3. **CVI Scotland**
4. **National Center on Deafblindness**
5. **Pediatric Cortical Visual Impairment Society (PCVIS)**
6. **Make it Easier Strategies**